

# Why Does My Baby Need More Testing for Congenital Adrenal Hyperplasia (CAH)?



State law requires that all babies have the newborn screening test before leaving the hospital. A few drops of blood were taken from your baby's heel and tested for certain diseases. Your baby now needs more testing as soon as possible. Not all babies with an initial "positive" result have CAH.

California  
Department of  
Health Services

Newborn Screening Program  
Genetic Disease Branch  
[www.dhs.ca.gov/gdb](http://www.dhs.ca.gov/gdb)



## Why Does My Baby Need More Testing?

Your baby's test showed a "positive" result for CAH due to 21-hydroxylase deficiency. More testing is needed to find out if your baby has this disease. Not all babies with an initial "positive" result will have this disease.

Babies can look healthy at birth and still have CAH. If this disease is not identified and treated quickly, serious health problems can occur.



## What Is CAH?

CAH is a disease where the adrenal glands are enlarged (hyperplasia) and do not work normally. The adrenal glands are small organs that are above the kidneys. Hormones are the body's messengers that tell the organs in the body what to do. A baby with CAH makes too much male hormone called androgen. This can lead to problems with growth and sexual development. Some babies have a more severe form and can not make enough of the hormone cortisol needed to maintain energy supply and blood sugar level. The baby may also not make enough of the hormone aldosterone. This causes too much salt and water to be lost in the urine. This can lead to dehydration.

## What Causes CAH?

CAH occurs when a certain enzyme is missing or not working right. Without this enzyme, little or no cortisol and aldosterone are made. Both of these hormones are needed to supply energy, maintain normal blood sugar level, and keep salt minerals in the body.

## How Does a Baby Get This Disease?

A baby with CAH inherited a gene from each parent. Both parents must carry the gene for a baby to inherit this disease.



## Is CAH Common?

About 35-50 babies a year in California are born with this disease.

## How Is This Disease Treated?

CAH is treated by giving the baby the missing hormones his/her body cannot make. The amount of medicine given will vary for each child. This amount will change as the child grows. Early treatment will help the child grow up healthy.



## What Happens Now?

The doctor will advise you as to what steps need to be taken. The NBS Program strongly recommends that newborns with positive screening test results be referred to a California Children's Services (CCS) approved Endocrine Special Care Center (SCC) or CCS paneled pediatric endocrinologist for a diagnostic evaluation including more testing.

## Who Will Pay for the Diagnostic Evaluation and Treatment If Needed?

All newborns referred to a CCS-approved Special Care Center (SCC) by the California Newborn Screening Program are eligible for a diagnostic evaluation through the SCC regardless of income. You will be asked to complete an application form to determine eligibility for CCS payment. Most health insurance and health maintenance organizations (HMOs) provide at least some coverage for the diagnostic evaluation and any necessary treatment. If your baby has health insurance, the SCC will bill the health insurance company or HMO for the services. Infants who have Medi-Cal full scope, no share of cost, or Healthy Families subscribers will be authorized by CCS for diagnostic and treatment services and parents will not need to pay anything for services. If you do not have health insurance, or if your insurance only covers partial payment, your infant may be eligible for the CCS program. To learn more about the CCS program visit their website at: <http://www.dhs.ca.gov/pcfh/cms/ccs/>.

## Is There Anything Else I Should Do?

Call your doctor **right away** if your baby:

- is not eating well
- has diarrhea or vomiting
- has any other signs of illness

For a complete list of diseases screened for by the California Newborn Screening Program, visit our website at [www.dhs.ca.gov/gdb](http://www.dhs.ca.gov/gdb)